Rare Diseases Take Spotlight in Annual Event

Patient groups, researchers, and health organizations are gearing up for the fourth annual Rare Disease Day, a global campaign that aims to raise awareness of the more than 250 million people worldwide who suffer from rare diseases.

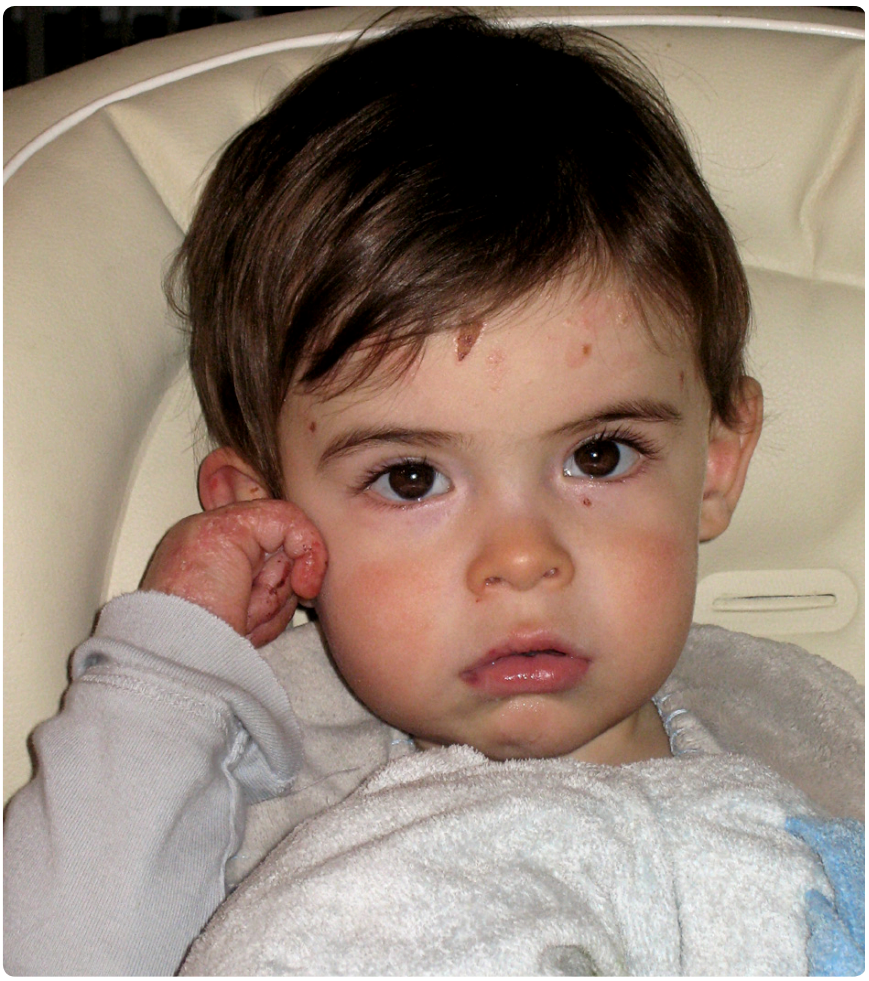
Held on the last day of February—Feb. 29 in leap years and Feb. 28 in other years—Rare Disease Day 2011 will focus on the disparities in access to services and treatment suffered by people with rare diseases.

About 7,000 known rare diseases have been identified around the world. Each disease has unique problems and may have little or no support or treatment. It is estimated that about 80 percent of rare diseases are genetic, and about half of all rare diseases affect children.

Rare disease is defined differently around the globe. In the United States, a disease is rare if it affects fewer than 200,000 people. About 30 million Americans are afflicted with rare diseases.

Among those hoping to raise awareness and find a cure for their own rare diseases are the following families who visited and shared their personal stories with the Food and Drug Administration's (FDA) Office of Orphan Products Development:

- Jackson has recessive dystrophic epidermolysis bullosa, a painful and incurable blistering disorder. In the hospital after his birth, an adhesive bandage put on his heel after a routine test ripped the skin from his foot when the bandage was removed. Jackson's tongue blistered and his mouth bled when he drank from a bottle. Jackson is now 3-1/2 years old. His hands, knees, heels, and elbows are always covered in bandages to prevent blisters, but Jackson still has scars from repeated blistering and continues to have difficulty eating. To learn more about Jackson and his disease, visit the Jackson Gabriel Silver Foundation (*www.jgsf.org*).
- Samantha (Sam) and Alexandria (Alex) have the rare progressive disease Friedreich's ataxia. This inherited disease of the nervous system results in difficulty moving and even speaking. Mary, the girls' mother, says there was no sign of a problem until Sam—then an active 9-year-



Jackson has recessive dystrophic epidermolysis bullosa, a painful and incurable blistering disorder. The 3-1/2 year old's hands, knees, heels, and elbows are often covered in bandages to help prevent blisters.

old—fell during a karate class. She was later given the diagnosis that changed her life. Sam's sister Alex was diagnosed several years later. Mary says their biggest challenge is that society looks at them as different. Despite this difficulty, Sam, 24, recently graduated from college and Alex, 20, is a junior in college. Both girls are very active and Mary calls them "kindness activists" in their efforts to raise awareness of Friedreich's ataxia while instilling the importance of kindness, acceptance, and dignity for all people. For more information about this disease, contact the Friedreich's Ataxia Research Alliance (*www.curefa.org*).

- Terry was diagnosed with mild Parkinson's disease in July 2006. As his symptoms rapidly got worse, he sought out another opinion and was then diagnosed at age 66 with progressive supranuclear palsy (PSP). Terry was an avid golfer and loved to dance with his wife, Ileen, but over the past several years he went from falling occasionally to using a cane, then a walker, and now a wheelchair. He cannot shower, dress himself, or sit up by himself without help. Terry says that when he was first diagnosed, he didn't know much about PSP and when he began to learn more he felt angry and hurt that he had this very debilitating disease. In spite of his challenges, Terry tries to remain active. He works out with a personal trainer two to three times a week and has acupuncture treatments, chiropractic care, and physical therapy, which help prevent stiffening of his body. For more information about this disease, contact CurePSP (*www.psp.org*).

FDA and Rare Diseases

FDA is in a unique position to help those who suffer from rare diseases. The agency's rare disease initiatives include the following:

- FDA's Office of Orphan Products Development gives grants to further the development of drugs, biologics (such as vaccines or blood products), medical devices, and medical foods for the treatment of rare diseases. (Medical foods are specially made and processed foods, used under a health care professional's supervision, to meet a patient's distinctive nutritional requirements.)
- FDA plays an important role in granting orphan designations and marketing approvals for drugs, biologics, and medical devices to treat rare diseases. Designating a potential product as "orphan" encourages companies to develop that product by giving them financial and other incentives.
- FDA's Office of New Drugs within the Center for Drug Evaluation and Research established the Rare Diseases Program to assist and support the research, development, regulation, and approval of drug and biologic products for the treatment of rare disorders.
- As countries combine their efforts to spotlight "Rare Diseases and Health Inequalities," FDA continues to collaborate with international regulatory agencies to address unmet medical needs of patients with rare diseases globally. FDA's Office of Orphan Products Development and the European Medicines Agency (EMA) work together regularly and have a common application for orphan product designation. (See EMA activities for Rare Disease Day at *www.ema.europa.eu*.)
- FDA advocates for rare disease awareness and treatments through work with patients and patient organizations.

U.S. Activities

The National Organization for Rare Disorders is sponsoring Rare Disease Day activities across the country. To find out what's going on in your area, visit Rare Disease Day USA (*www.rarediseaseday.us*).

In the Washington, D.C., area, the National Institutes of Health (NIH) and FDA's Office of Orphan Products Development are participating in a day-long celebration and recognition of the various rare diseases research activities at NIH's Lipsett Amphitheater. For an agenda of the day's activities, click here (*www.rarediseases.info.nih.gov/files/Rare Dz Day agenda.pdf*). Activities begin at 8:30 a.m. and conclude at 5:15 p.m.

Attendance at the NIH event is free and open to the public, but advance registration (*rarediseases.info.nih.gov/Rare_Disease_Day/AddContact.aspx*) is encouraged. Visitors should check the NIH Visitors and Security Web page (*www.nih.gov/about/visitorsecurity.htm*) for instructions and allow plenty of time to pass through security. In support of the Global Genes Project—a rare disease support-building and fundraising campaign—all attendees are encouraged to wear their favorite pair of jeans.

Sign language interpreters will be provided. People with disabilities who need reasonable accommodation to participate may contact Kimberly Potter (*kpotter@icfi.com*) at 301-251-4962 or the Federal TTY Relay number at 1-800-877-8339.

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